



sensorineural deafness and male infertility

Sensorineural deafness and male infertility is a condition characterized by hearing loss and an inability to father children. Affected individuals have moderate to severe sensorineural hearing loss, which is caused by abnormalities in the inner ear. The hearing loss is typically diagnosed in early childhood and does not worsen over time. Males with this condition produce sperm that have decreased movement (motility), causing affected males to be infertile.

Frequency

The prevalence of sensorineural deafness and male infertility is unknown.

Genetic Changes

Sensorineural deafness and male infertility is caused by a deletion of genetic material on the long (q) arm of chromosome 15. The signs and symptoms of sensorineural deafness and male infertility are related to the loss of multiple genes in this region. The size of the deletion varies among affected individuals. Researchers have determined that the loss of a particular gene on chromosome 15, the *STRC* gene, is responsible for hearing loss in affected individuals. The loss of another gene, *CATSPER2*, in the same region of chromosome 15 is responsible for the sperm abnormalities and infertility in affected males. Researchers are working to determine how the loss of additional genes in the deleted region affects people with sensorineural deafness and male infertility.

Inheritance Pattern

Sensorineural deafness and male infertility is inherited in an autosomal recessive pattern, which means both copies of chromosome 15 in each cell have a deletion. The parents of an individual with sensorineural deafness and male infertility each carry one copy of the chromosome 15 deletion, but they do not show symptoms of the condition.

Males with two chromosome 15 deletions in each cell have sensorineural deafness and infertility. Females with two chromosome 15 deletions in each cell have sensorineural deafness as their only symptom because the *CATSPER2* gene deletions affect sperm function, and women do not produce sperm.

Other Names for This Condition

- chromosome 15q15.3 deletion syndrome
- deafness-infertility syndrome
- DIS

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Deafness-infertility syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1970187/>

Other Diagnosis and Management Resources

- Cleveland Clinic: Male Infertility
<http://my.clevelandclinic.org/health/articles/male-infertility>
- GeneReview: CATSPER-Related Male Infertility
<https://www.ncbi.nlm.nih.gov/books/NBK22925>
- MedlinePlus Health Topic: Assisted Reproductive Technology
<https://medlineplus.gov/assistedreproductivetechnology.html>
- RESOLVE: The National Infertility Association: Semen Analysis
<http://www.resolve.org/about-infertility/male-workup/the-semen-analysis.html>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Sensorineural Deafness
<https://medlineplus.gov/ency/article/003291.htm>
- Health Topic: Assisted Reproductive Technology
<https://medlineplus.gov/assistedreproductivetechnology.html>
- Health Topic: Hearing Disorders and Deafness
<https://medlineplus.gov/hearingdisordersanddeafness.html>
- Health Topic: Male Infertility
<https://medlineplus.gov/maleinfertility.html>

Genetic and Rare Diseases Information Center

- Deafness-infertility syndrome
<https://rarediseases.info.nih.gov/diseases/11911/deafness-infertility-syndrome>

Educational Resources

- Boston Children's Hospital: Sensorineural Hearing Loss
<http://www.childrenshospital.org/conditions-and-treatments/conditions/sensorineural-hearing-loss>
- Boys Town National Research Hospital: The Normal Ear
<https://www.boystownhospital.org/knowledgeCenter/articles/hearing/Pages/TheNormalEar.aspx>
- Cleveland Clinic: Male Infertility
<http://my.clevelandclinic.org/health/articles/male-infertility>
- Disease InfoSearch: Deafness, sensorineural, and male infertility
<http://www.diseaseinfosearch.org/Deafness%2C+sensorineural%2C+and+male+infertility/8194>
- MalaCards: deafness-infertility syndrome
http://www.malacards.org/card/deafness_infertility_syndrome
- Merck Manual Professional Version: Sperm Disorders
<http://www.merckmanuals.com/professional/gynecology-and-obstetrics/infertility/sperm-disorders>
- Orphanet: Deafness-infertility syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=94064

Patient Support and Advocacy Resources

- RESOLVE: The National Infertility Association
<http://www.resolve.org/>
- University of Kansas Medical Center Resource List: Hard of Hearing/Deafness
<http://www.kumc.edu/gec/support/hearing.html>
- University of Kansas Medical Center Resource List: Infertility
<http://www.kumc.edu/gec/support/inferti.html>

GeneReviews

- CATSPER-Related Male Infertility
<https://www.ncbi.nlm.nih.gov/books/NBK22925>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22Infertility%2C+Male%22+OR+%22sensorineural+deafness+and+male+infertility%22+OR+%22Male+Infertility%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28sensorineural+deafness+and+male+infertility%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- DEAFNESS-INFERTILITY SYNDROME
<http://omim.org/entry/611102>

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